ADOPT BBMRI-ERIC
GRANT AGREEMENT NO. 676550

DELIVERABLE REPORT

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<td>Deliverable Title</td>
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BEST PRACTICE DOCUMENT FOR OPTIMAL USAGE OF OMICS TECHNOLOGIES FOR BIOMARKERS

Executive Summary

Different omics technologies are expected to accelerate biomarker discovery and validation but translation of these technologies into clinically actionable tools has been slow. Multiple technical platforms exist for omics profiling and there was a need to survey the operational and quality aspects of the omics technology platforms. With the help of the National Nodes, we produced a survey of the omics technology platforms in BBMRI-ERIC Member States. We received information from a total of 54 omics service providers from twelve BBMRI-ERIC Member States. Most of the results were obtained from genomics or transcriptomics platforms where the used technologies are already well validated. In the areas of metabolomics and proteomics the validation of technologies is still ongoing. The results of the questionnaires indicated that most of the omics service platforms are operating under quality control procedures but the quality of samples delivered by biobanks or researchers was generally not followed. The BBMRI-ERIC associated biobanks could clearly take a prominent position in jointly implementing international ISO and CEN/TS standards applicable for samples intended for omics analyses. Further collaboration with the technology platforms should be encouraged. The current results of the omics survey can support the biobankers and researchers to find omics technology platforms that have open access, operate under defined quality conditions and have...
been utilized to analyze high quality samples of BBMRI-ERIC biobanks or researchers utilizing biobank samples.

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This project has received funding from the European Union’s Horizon 2020 research and innovation programme under grant agreement No 676550.

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1. Background

Omics technologies such as genomics, transcriptomics, metabolomics and proteomics enable simultaneous and holistic measurement of genes, gene expression, proteins and metabolites and these technologies are expected to accelerate the rate of biomarker discovery and validation. The promise of omics technologies is considered huge, but translation of these technologies into clinically actionable tools has been quite slow. Multiple technical platforms from different manufacturers are available for omics profiling and it is not always clear to the user which technology platform would give reliable results. Also, for biobanks it is essential to know those technology platforms that are able to return quality defined analysis data back to biobanks.

The fact is that standardized and harmonized protocols are still lacking within the omics field. Validation of omics technologies is far from complete and best practices for performing omics studies are yet to be established. Biobanks can participate in the development of best practices for omics studies because defined preanalytical conditions are a prerequisite for reproducible results. There is an increasing awareness of the need for high quality biospecimens and implementing QC tools assuring reliable results in biomarker studies.

More information of required sample quality and also quality of the technology providers is needed to reach the full potential of utilization of biobank samples in omics technologies. In this task we collaborated with the BBMRI-ERIC National Nodes to obtain information of omics technology platforms in BBMRI-ERIC Member States. The aim was to establish initial quality criteria and best practices for omics specific sampling for genomics, transcriptomics, metabolomics and proteomics.

With the help of the National Nodes, we produced a survey of the omics technology platforms in BBMRI-ERIC Member States. Furthermore, we collected here complementing information from the BBMRI-LPC project and other BBMRI initiatives.

2. Approaches (Methods)

The planning of the task was initiated in autumn of 2016. The topics for the questionnaires were discussed at the BBMRI-ERIC Management Committee (MC) meeting and a core working group, consisting of National Node directors from BBMRI.fi, BBMRI.at, BBMRI.nl and BBMRI.it was agreed and the omics questionnaire was planned in collaboration with ADOPT BBMRI-ERIC WP2. The questionnaire was created during May and June 2017. This included teleconferences between the core working group and consulting of 6 Finnish experts in omics analyses and technology platforms. The completed questionnaire was sent to the directors of BBMRI National Nodes in July 2017 with a cover letter, where they were asked to circulate the questionnaire to all relevant technology platforms in their country. Also, the BARC-database produced by BBMRI.se and BBMRI-LPC was used to identify some of the relevant actors, which were contacted directly. The collection of data proceeded relatively slowly and the results of the questionnaire were presented and further discussed in the ADOPT project meeting of (Wrocław,
October 10th 2017). After the Wroclaw meeting the National Node directors took one more effort to get more answers to the questionnaires and we actually could almost double the amount of data. The answers to the questionnaires were received between July and early November and analyzed in November 2017.

3. Results
In total we received 54 answers to the 4 questionnaires. We received
- 23 answers to the genomics-questionnaire,
- 14 to the transcriptomics-questionnaire,
- 6 answers to the metabolomics-questionnaire
- 11 answers to the proteomics-questionnaire,

from omics technology platforms located in twelve BBMRI-ERIC Member States:
- Austria
- Estonia
- Finland
- France
- Germany
- Italy
- Latvia
- Malta
- Poland
- Sweden
- The Netherlands
- Turkey

The data collected in the questionnaires can be found in Appendix 2 a-d.

3.1 Genomics
The majority (16/23) of the institutions answering genomics-questionnaire reported to have open access. Majority of the institutions have determined required quality standards for the samples and the facility operates under strict quality control procedures. The most common quality standards for the facilities were ISO17025 and ISO9001, but also others were used (i.e; in France the French national norm for technological platform in research NFX 50-900). The quality standards required from the samples were more scattered and this is clearly an area where BBMRI-ERIC biobanks could have an impact. The quantity of required DNA varied from 50ng to 4µg. The required DNA concentrations varied between 2ng/µl to 60ng/µl, and most frequently 50ng/µl was required. Almost every facility was performing quality control to the samples, either themselves or through a close collaborator and almost everyone used Illumina as at least one of their technologies. Also raw files were delivered by the majority of the facilities, with other deliverables varying from comprehensive to none. It seems that the analysis technologies are well established but there is a need to agree on preanalytical standards of DNA samples and
also the analysis and standardization of the results currently needs major input from the scientists of the receiving institute.

### 3.2 Transcriptomics

Most of the institutions answering the transcriptomics-questionnaire also reported to have open access (11/14). 7/14 respondents reported to have a quality standard for their facilities. Only one of the facilities had a ISO standard for the samples, while others required for example the RNA Integrity (RIN)- value to fall between 5-10. The quantity of required RNA needed for IVT reactions ranged from 1ng up to 1,5µg. The required concentrations varied between 10ng/µl to 500ng/µl. All facilities but one were performing quality control for their operations.

### 3.3 Metabolomics

Metabolomics services are currently produced only by a few technology platforms. We obtained data only from six platforms and two of them reported to have open access and being certified either for ISO 13485 or ISO 17025. One of the facilities was using NMR while others were doing MS-metabolomics. The required quantity of the serum/plasma was between 5-500µl. Every facility was doing quality control and the NMR platform has also initiated validation of their methodology with a clear aim in producing clinically relevant analysis tools.

### 3.4 Proteomics

We obtained data from 11 platforms and seven of them reported to have open access. Six of them had an official quality standard for the facility, all of which were using at least ISO9001. Only one of the facilities reported not to provide the quality control. The used technologies and throughput of deliverables varied largely between the facilities.

### 3.5 Omics related information collected in other initiatives

- BARC-database produced by BBMRI.se and BBMRI-LPC\(^1\)
- BBMRI.nl has collected information from omics data collected in international collaboration activities. This data is found in the BBMRI-OMICS database\(^2\). It consists of omics data that has been generated and that is made available for BBMRI researchers focusing on integrative omics studies in Dutch Biobanks
- Core Technology for the Life Science network has technology platforms as members and more information on the omics technology platforms can be searched through the network web pages\(^3\).

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This project has received funding from the European Union’s Horizon 2020 research and innovation programme under grant agreement No 676550.
3.6 BBMRI-ERIC operates actively towards biobank quality procedures

- Implementation of international ISO and CEN/TS applicable for biobanks
- BBMRI-ERIC will organize a self-evaluation of biobanks during 2017
- The ADOPT BBMRI-ERIC IT tool and Self-Assessment Survey (D6.2) supports the structured monitoring of the pre-analytical process of samples as defined in the CEN/TS. This tool also provides key data for monitoring interoperability and quality of biobanks and appropriate collections.

4. Discussion and Conclusions

4.1 Towards best practices for optimal usage of omics technologies

The original aim of this survey was to produce a best practice document for the utilization of omics technologies for biobanks and researchers. The collection of initial information from the omics platforms was slow and as a result, a deeper analysis towards best practices could not be delivered within the ADOPT.

The current results of the omics survey can support the biobankers and researchers to find omics technology platforms that have open access, operate under defined quality conditions and have been utilized to analyze high quality samples of BBMRI-ERIC biobanks or researchers utilizing biobank samples.

Requirements for sample processing

- Biobanks should widely implement international ISO and CEN/TS standards applicable for samples to be delivered for omics analyses
- Minimum requirement for the standard monitoring of pre-analytical processes of samples is advised to be monitored by using the ADOPT BBMRI-ERIC IT tool and Self-Assessment Survey

Minimal requirements for the omics technology platform

- Technology platforms with open access should be preferred
- Technology platform should implement ISO quality standards for the platform
- A well-established technology platform performs a quality check of the imported samples. As a result, a double quality check for the samples is preferred

Test validation and documentation

- The omics technology platform should document all methods and procedures
5. Next Steps

Discussions have been initiated towards collaboration with the BARC-database and BBMRI-LPC project participants to explore the possibilities to incorporate collected information with BARC-database. It would be important to identify a BBMRI-ERIC National node that would have interest in hosting the database of the omics technology platforms.

Close contacts and continuous discussion between biobanks and technology platforms will be necessary to further develop the awareness of the sample quality issues and to jointly develop the analytics. The coordination of such activities remains to be organized between BBMRI-ERIC headquarters and the National Nodes.
6. References

1 BARC-database: www.barcdb.org
2 BBMRI-OMICS database: www.health-ri.org/services
3 Core technology for the Life Science (CTLS) network: www.ctls-org.eu
4 European Committee for Standardisation - cen.eu
5 BBMRI-ERIC Quality Policy - www.bbmri-eric.eu/services/standardisation/
7. Appendices

Appendix I: List of omics technology platforms
   a) Based on genomics-questionnaire
   b) Based on transcriptomics-questionnaire
   c) Based on metabolomics-questionnaire
   d) Based on proteomics-questionnaire

Appendix II: a) Table of results for the genomics-questionnaire
   b) Table of results for the transcriptomics-questionnaire
   c) Table of results for the metabolomics-questionnaire
   d) Table of results for the proteomics-questionnaire

Appendix III: Abbreviations

Appendix IV: Questionnaires
   a) Genomics-questionnaire
   b) Transcriptomics-questionnaire
   c) Metabolomics-questionnaire
   d) Proteomics-questionnaire

Appendix I: List of omics technology platforms

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<th>Name</th>
<th>E-mail</th>
<th>Phone number</th>
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<td>The Estonian Genome Center Core Facility</td>
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---

**b) Based on transcriptomics-questionnaire**

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<th>Name</th>
<th>E-mail</th>
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This project has received funding from the European Union’s Horizon 2020 research and innovation programme under grant agreement No 676550.
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c) Based on metabolomics-questionnaire

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d) Based on proteomics-questionnaire

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## Appendix II: Tables of results

### a) Table of results for the genomics-questionnaire

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<th>Has open access</th>
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<th>Raw files inc. IDAT files</th>
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<td></td>
</tr>
<tr>
<td>Helmholtz Zentrum München - GAC</td>
<td>DE</td>
<td>Yes</td>
<td>ISO</td>
<td>non-degraded genomic DNA</td>
<td>500 ng genomic DNA</td>
<td>60 ng/µl</td>
<td>Yes</td>
<td>Affymetrix, Illumina, Agena</td>
<td>5000 samples / month</td>
<td>x</td>
<td></td>
<td>x  x</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Biobank Lab, University of Lodz</td>
<td>PL</td>
<td>Yes</td>
<td>In progress</td>
<td>Some IBBL PT</td>
<td>Depends on technique</td>
<td>Depends on platform - for arrays 50ng/mL</td>
<td>Yes</td>
<td>Illumina</td>
<td>800 ma/week, up to 200 small genomes</td>
<td>x  x x x</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>FIMM genotyping unit</td>
<td>FI</td>
<td>Yes</td>
<td>None</td>
<td>Depends on who is sending us the samples</td>
<td>Yes</td>
<td>Depends on the technology</td>
<td>No</td>
<td>Affymetrix, Illumina, Agena</td>
<td>Depends on which technology is used</td>
<td>x  x x x</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>The SNP&amp;SEQ Technology Platform</td>
<td>SE</td>
<td>Yes</td>
<td>ISO17025:2005</td>
<td>ISO17025:2005</td>
<td>200-400ng</td>
<td>min.15ng/µl, recommended 50ng/µl</td>
<td>Yes</td>
<td>Illumina</td>
<td>35K smpls/yr for SNP genotyping</td>
<td>x  x x x</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Genome Database of Latvian population</td>
<td>LV</td>
<td>Yes</td>
<td>none at the moment</td>
<td>Compliant with CEN/TS 16835-2</td>
<td>500 ng</td>
<td>over 50 ng/ul</td>
<td>Yes</td>
<td>Ion Torrent, Ion Proton</td>
<td>110 runs per year</td>
<td>x</td>
<td>x</td>
<td>x</td>
<td></td>
<td></td>
</tr>
<tr>
<td>--------------------------------------</td>
<td>----</td>
<td>-----</td>
<td>-------------------</td>
<td>-------------------------------</td>
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<td>-----</td>
<td>------------------------</td>
<td>------------------</td>
<td>---</td>
<td>---</td>
<td>---</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Bioinformatics Long-term Support</td>
<td>SE</td>
<td>Yes</td>
<td>None</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>No</td>
<td>Knoe-how in bioinfo omics (only staff)</td>
<td>25 large project per year</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clinical Genomics Uppsala</td>
<td>SE</td>
<td>Yes</td>
<td>Some diagnostic tests are ISO15189</td>
<td>Genomics Facility (not biobank)</td>
<td>Highly dependent on method</td>
<td>Highly dependent on method</td>
<td>Yes</td>
<td>Illumina, NGS using several technologies,</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clinical Genomics Stockholm (SciLifeLab)</td>
<td>SE</td>
<td>No</td>
<td>ISO 17025</td>
<td>-</td>
<td>variable</td>
<td>variable</td>
<td>Yes</td>
<td>Illumina</td>
<td>10000 samples</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>INT Biobank</td>
<td>IT</td>
<td>only for collaborative studies</td>
<td>ISO9001</td>
<td>not clear for me this query</td>
<td>50-500 ng</td>
<td>2-50 ng/microl</td>
<td>Yes</td>
<td>Affymetrix, Illumina, Agilent, Thermo Fisher</td>
<td>650/year</td>
<td>x</td>
<td>x</td>
<td>x</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Leiden Genome Technology Center (LGTC)</td>
<td>NL</td>
<td>Yes</td>
<td>N/A</td>
<td>N/A</td>
<td>Depends on experiment</td>
<td>Depends on experiment</td>
<td>Yes</td>
<td>Illumina, PacBio, Bionano</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Ankara University Biotechnology Institute</td>
<td>TR</td>
<td>Yes</td>
<td>GLP is followed</td>
<td>DNA checked by gel electrophoresisOD260/OD 280=1.75-1.85</td>
<td>312.5 ng per 250K SNP array</td>
<td>50 ng/ul</td>
<td>Yes</td>
<td>Affymetrix</td>
<td>30-50/month</td>
<td>x</td>
<td></td>
<td>x</td>
<td></td>
<td></td>
</tr>
<tr>
<td>GenomeScan BV</td>
<td>NL</td>
<td>NO</td>
<td>ISO 17025/GLP</td>
<td>various</td>
<td>various</td>
<td>various</td>
<td>Yes</td>
<td>Affymetrix, Illumina, PacBio/ LifeTech</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

This project has received funding from the European Union’s Horizon 2020 research and innovation programme under grant agreement No 676550.
| IBVR, IZSLER Biobank of veterinary resources | IT | no | ISO 17025 | purified dsDNA | 300 ng | up to 10 ng/ul | Yes | Illumina | 200 samples/year | x |
| BaseClear bv. | NL | yes | ISO 17025 | ? | We can work with very low quantities | We can work with very low quantities | Yes | Illumina, PacBio; Oxford Nanopore | Hundreds per week | x | x | x | x |
| HUNT Biobank | NO | Yes | ISO 9001 | ISO 9001 | 200 ng | 25-50 ng/ul | Yes | Illumina | 4000 chiparrays/week | x | x | x | x | x |
| Microsynth Austria | AT | no | ISO 9001: 2015, ISO/IEC 17025: 2005, GMP | we have no sample collection | 0.5 µg | 10 ng/µl | Yes | Illumina, Sanger Sequencing | x |
| INRA Gentyane | FR | yes | ISO 9001: 2015: NF X 50-900 | high | 100 ng - 10µg | 10 - 50 ng/µl | Yes | Affymetrix, Illumina, Pacific Bioscience | 8 chips Affymetrix/week, 14 runs Sequel PacBio/week | x | x | x | x |
| FLUXGEN | FR | no | ISO 9001 | cDNA ozyme | 3 µL | 5ng/mL | Yes | fluidigm biomark HD | x |
| NTNU Genomics Core Facility | NO | Yes | Illumina CSpRo | - | - | - | Yes | Illumina | x | x | x |
| Translational Research | FR | ips2.u-psud.fr | IBISA & CNOC (national standards) | Not relevant | Not relevant | Not relevant | Yes | Targeting Induced Local Lesions in Genomes | 30 genes/month |
| Ligan-MP | FR | access via services & collab. | no | ratio 260/280: >1,8 | 500 ng | 50ng/µl | No | Illumina | 96 arrays/week | x | x | x | x | x |
| MAD: Dutch Genomics Service & Support Provider | NL | Yes | Experience | Provided by customers | Depends on the application | Depends on the application | Yes | Affymetrix, NGS: Ion Proton and OxfordNano por | > 60 small to large microarray/NGS projects | x | x | x | x |

This project has received funding from the European Union’s Horizon 2020 research and innovation programme under grant agreement No 676550.
b) Table of results for the transcriptomics-questionnaire

<table>
<thead>
<tr>
<th>Name of facility</th>
<th>Country</th>
<th>Facility has open access</th>
<th>Quality standard of facility</th>
<th>Quality standard of samples</th>
<th>Quantity of RNA required</th>
<th>Concentration of RNA required</th>
<th>QC provided by the facility</th>
<th>Used technology</th>
<th>Throughput capacity</th>
<th>Deliverable 1</th>
<th>Deliverable 2</th>
<th>Deliverable 3</th>
<th>Deliverable 4</th>
</tr>
</thead>
<tbody>
<tr>
<td>Estonian Genome Center Core Facility</td>
<td>EE</td>
<td>yes</td>
<td>ISO 9001:2008</td>
<td>ISO 9001:2008</td>
<td>minimum 0,5 ug</td>
<td>50 ng/ul</td>
<td>Yes</td>
<td>Illumina</td>
<td>2000</td>
<td>raw data</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Genome Database of Latvian population</td>
<td>LV</td>
<td>Yes</td>
<td>none at the moment</td>
<td>RIN nuber over 6</td>
<td>over 100 ng</td>
<td>10 ng/ul</td>
<td>Yes</td>
<td>Ion Torrent, Ion Proton</td>
<td>110 runs per year</td>
<td>Raw files</td>
<td>FASTQ files</td>
<td></td>
<td>Differentially expressed gene list</td>
</tr>
<tr>
<td>INT Biobank</td>
<td>IT</td>
<td>only for collaborative studies</td>
<td>ISO9001</td>
<td>unclear query for me</td>
<td>50 ng- 1 µg</td>
<td>10 - 100 ng/ microl</td>
<td>Yes</td>
<td>Affymetrix, Illumina, ThermoFisher, Agilent, QuantStudio 12K Flex</td>
<td>2,500/year</td>
<td>BAN FASTQ files</td>
<td>CEL files</td>
<td></td>
<td>TXT files</td>
</tr>
<tr>
<td>Ankara University Bio technology Institute</td>
<td>TR</td>
<td>Yes</td>
<td>GLP is followed though no certification for ISO etc.</td>
<td>RNA integrity by Bioanalyzer (min RIN 7.5), OD260/ OD280=1.9-2</td>
<td>1.5 µg for IVT reactions</td>
<td>500 ng/ul</td>
<td>Yes</td>
<td>Affymetrix</td>
<td>30-50/month</td>
<td>CEL</td>
<td>ChP</td>
<td>Differentially expressed gene list</td>
<td>Cluster, Pathway and Ontology analyses results</td>
</tr>
<tr>
<td>SCIBLU Genomics</td>
<td>SE</td>
<td>Yes</td>
<td>No</td>
<td>Pure RNA,RIN as close to 10 as possible.</td>
<td>100-200 ng</td>
<td>ca 100 ng/µl</td>
<td>Yes</td>
<td>Affymetrix</td>
<td>usually ca 1000 samples per year</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Microsynth Austria</td>
<td>AT</td>
<td>no</td>
<td>ISO 9001:2015, ISO/IEC 17025:2005</td>
<td>we have no sample collection</td>
<td>e1µg</td>
<td>e20ng/µl</td>
<td>Yes</td>
<td>Illumina</td>
<td>-</td>
<td>5 Mio reads, 1<em>75 or 1</em>150</td>
<td>10 Mio reads, 1<em>75 or 1</em>150</td>
<td>30 Mio reads, 1<em>75 or 1</em>150</td>
<td>50 Mio reads, 1<em>75 or 1</em>150</td>
</tr>
<tr>
<td>IBENS Genomics facility</td>
<td>FR</td>
<td>Yes</td>
<td>ISO 9001 NFX 50-900</td>
<td>Verified by the facility (qubit, bioanalyzer)</td>
<td>10 ng min.</td>
<td>200 ng/µL</td>
<td>Yes</td>
<td>Illumina</td>
<td>70 project and 800 samples / year</td>
<td>Fastq files</td>
<td>FastQC and FastqScreen reports</td>
<td>Read alignments and counts</td>
<td>Differential analysis</td>
</tr>
</tbody>
</table>

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<table>
<thead>
<tr>
<th>GenomEast Platform</th>
<th>FR</th>
<th>yes</th>
<th>ISO9001, NFX50-900</th>
<th>NA</th>
<th>from 1 ng to 1 µg</th>
<th>Not required</th>
<th>Yes</th>
<th>Illumina</th>
<th>3500 samples per year</th>
<th>FASTQ files</th>
<th>md5 of FASTQ files</th>
<th>Samples and libraries quality control results</th>
<th>Sequencing quality control reports</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genomic Paris Centre iBENS</td>
<td>FR</td>
<td>yes</td>
<td>ISO 9001; NFX 50-900</td>
<td>more than 5 if possible</td>
<td>min 1 ng to begin</td>
<td>200 ng/µl if possible</td>
<td>Yes</td>
<td>Illumina, Oxford Nanopore Technologies</td>
<td>-</td>
<td>fastq and fastqc</td>
<td>filtered alignment files (BAM and BAI formats), files for genome viewers (bedGraph)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>UCA GenomiX</td>
<td>FR</td>
<td>yes</td>
<td>ISO9001</td>
<td>very good</td>
<td>100 ng</td>
<td>1</td>
<td>Yes</td>
<td>Illumina, 10x Genomics</td>
<td>1Tb/month</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>NTNU Genomics Core Facility</td>
<td>NO</td>
<td>Yes</td>
<td>Illumina CSPro</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>Yes</td>
<td>Illumina</td>
<td>-</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Ligan-MP</td>
<td>FR</td>
<td>access via services and collaborations</td>
<td>Not yet. We plan to work using ISO 15189</td>
<td>RINs for each sample by the customer</td>
<td>to be discussed</td>
<td>depending on each project</td>
<td>No</td>
<td>Illumina, RNA sequencing</td>
<td>FastQ data</td>
<td>counts</td>
<td>other on demande</td>
<td></td>
<td></td>
</tr>
<tr>
<td>MAD: Dutch Genomics Service &amp; Support Provider</td>
<td>NL</td>
<td>yes</td>
<td>Experience</td>
<td>provided by customer</td>
<td>Depends on the application</td>
<td>Depends on the application</td>
<td>Yes</td>
<td>Affymetrix, I-Proton-small-RNA-seq</td>
<td>&gt;60 small &amp; large projects per year</td>
<td>Anything a customer demands...</td>
<td>Lots of specialized bioinformatics</td>
<td></td>
<td></td>
</tr>
<tr>
<td>platform &quot;Exploration of Metabolism&quot;, transcriptomic team</td>
<td>FR</td>
<td>Yes</td>
<td>internal quality standard</td>
<td>MIQE</td>
<td>1 µg</td>
<td>25 ng/µl</td>
<td>Yes</td>
<td>Agilent microarrays - Applied Biosystems (q PCR)</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

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### Table of results for the metabolomics-questionnaire

<table>
<thead>
<tr>
<th>Name of facility</th>
<th>Country</th>
<th>Facility has open access</th>
<th>Quality standard of facility</th>
<th>Quality standard of samples</th>
<th>Quantity of serum/plasma required</th>
<th>QC provided by the facility</th>
<th>Used technology</th>
<th>Throughput capacity</th>
<th>Deliverable 1</th>
<th>Deliverable 2</th>
<th>Deliverable 3</th>
<th>Deliverable 4</th>
<th>Deliverable 5</th>
</tr>
</thead>
<tbody>
<tr>
<td>Helmholtz Zentrum München, Genome Analysis Center</td>
<td>DE</td>
<td>No</td>
<td>research</td>
<td>research</td>
<td>depending on the assay (50-500 µL)</td>
<td>Yes</td>
<td>MS-metabolomics</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Metanomics Health GmbH</td>
<td>DE</td>
<td>No</td>
<td>Proprietary Work Instructions</td>
<td>SOPs</td>
<td>50µL per sample</td>
<td>Yes</td>
<td>MS-metabolomics</td>
<td>Sample type check</td>
<td>Blood Processing Control</td>
<td>Coagulation Quality Control</td>
<td>Sample Processing Control</td>
<td>Overall Quality</td>
<td></td>
</tr>
<tr>
<td>Metabolomics Unit, FIMM, HiLIFE, UH.</td>
<td>FI</td>
<td>Yes</td>
<td>Internal QC</td>
<td>Checked</td>
<td>100 µL</td>
<td>Yes</td>
<td>MS-metabolomics</td>
<td>2000 samples per year</td>
<td>Few weeks</td>
<td>Report in Word format</td>
<td>Data in Excel format</td>
<td>Results in ppt format</td>
<td>Manuscript writing</td>
</tr>
<tr>
<td>Nightingale Health Ltd</td>
<td>FI</td>
<td>No</td>
<td>EN ISO 13485</td>
<td>high</td>
<td>100µL</td>
<td>Yes</td>
<td>NMR</td>
<td>40000</td>
<td>228 metabolic biomarkers</td>
<td>Absolute quantified metabolites</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>ICANalytics</td>
<td>FR</td>
<td>Yes</td>
<td>ISO 9001 (in progress)</td>
<td>French standard - NF S96-900 (in progress)</td>
<td>50-250 µl depends on LC-MS method</td>
<td>Yes</td>
<td>MS-metabolomics</td>
<td>4000 samples per year</td>
<td>Excel file - DataMATRIX</td>
<td>.pdf Report</td>
<td>.ppt and meeting</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Biomedical Metabolomics Facility Leiden</td>
<td>NL</td>
<td>users do not operate instruments</td>
<td>ISO 17-025</td>
<td>study pool is used as QC</td>
<td>from 5 µL to 500µL depending on target compounds</td>
<td>Yes</td>
<td>MS-metabolomics</td>
<td>15000 per year</td>
<td>validated targeted platforms</td>
<td>global profiling</td>
<td>miniaturised, 3D cell culture</td>
<td>lipidomics</td>
<td></td>
</tr>
</tbody>
</table>
**d) Table of results for the proteomics-questionnaire**

| Name of facility                                | Country | Facility has open access | Quality standard of facility | Quality standard of samples | Quantity of serum/plasma required | QC provided by the facility | Tech 1                                                                 | Tech 2                                                                 | Tech 3                                                                 | Tech 4                                                                 | Throughput capacity                                                                 | Deliverable 1 | Deliverable 2 | Deliverable 3 | Deliverable 4 | Deliverable 5 |
|------------------------------------------------|---------|--------------------------|------------------------------|-----------------------------|---------------------------------|-------------------------------|----------------------------|------------------------------------------------|------------------------------------------------|------------------------------------------------|------------------------------------------------|------------------------------------------------|----------------|----------------|----------------|----------------|----------------|
| Meilahti Clinical Proteomics Core Unit          | FI      | yes                      | GLP                          | GLP                         | micrograms                      | Yes                           | clinical proteomics, MRM, PRM, Imaging MS | Glyco-proteomics, carbohydrate analysis and identification | systems proteomics, -medicine and - biology | DIGE, 1D and 2D analysis, micro and nano UHPLC | 120 clin prot,P/M, 120 glyco prot,P/M, 200-400 IMS P/M | 1200 samples/year | ISO:9001-2015 | NLX-50-900 | NLX-50-900 | NLX-50-900 |
| Proteomics @PSL (SMBP-ESPCI)                     | FR      | yes                      | N/A                          | N/A                         | 100 µL                          | Yes                           | MALDI TOF/TOF | nanoESI FTICR | nanoESI Q Exactive (HF) | nanoESI LTQ Orbitrap | 800-800/month in LCMS, more in MS only | identification of protein by peptide sequencing | stable isotope or label free quantification | redox state of proteins (oxidation, nitrosylation) | whole protein mass analysis | interactome and protein complexes study |
| PP2I                                            | FR      | yes                      | ISO9001                      | NO                          | Yes                             | Depending of the type of analyse | MALDI TOF | ESI TRIPLE TOF | CESI               | 60 TO 100 ANALYSES PER MONTH | feasibility with our equipments | sensitivity required | sample processing | protein identification and/or quantification |
| Functional Proteomics Platform                   | FR      | yes                      | ISO9001-2015                 | 10 µg                       | Yes                             | Guidelines of high ranked journals | nanoLC-FT-MS/MS | NanoESI-FT-MS/MS | Electrophoresis | 1000 samples/month | protein identification | Post-translational modifications | N-terminal sequence | peptide sequencing |
| PISSARO                                         | FR      | yes                      | ISO 9001, NFX-50-900         | none                        | few µL                          | Yes                           | mass spectrometry | Chromatography | N-terminal micro-sequencing | Electrophoresis | protein identification | Post-translational modifications | N-terminal sequence | peptide sequencing |
| Center for Proteomics and Metabolomics at the Leiden University Medical Center | NL      | no                       | none                        | -80 and -20 sample storage, dependent technology applied | 2 µl to 100 µl | Yes | Mass spectrometry | glycomics of serum or plasma | NMR metabolomics platform by LC-MS | targeted lipidomics platform by LC-MS | up to 2000 samples per month, depends on technology |
| PIOTM                                           | FR      | yes                      | ISO 9001 v2015 & NF X50-900v2016 | No                          | 1 mL                            | Yes                           | shotgun proteomics | label free differential proteomics | post-translational modification analysis | mass spectrometry imaging | 700 samples per year | N/A | N/A | N/A | N/A | N/A |

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<table>
<thead>
<tr>
<th>Location</th>
<th>Country</th>
<th>ISO9001</th>
<th>200 µL - Less to be tested</th>
<th>NanolC-MS/MS ESI-Trap</th>
<th>MALDI-TOF/TOF</th>
<th>High resol nanoLC-MS/MS soon available</th>
<th>1000 / year</th>
<th>Turnaround time small-scale experiments</th>
<th>Turnaround time large-scale experiments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Plateforme P3S - UMS omique</td>
<td>FR</td>
<td>Yes</td>
<td>none</td>
<td>Yes</td>
<td>nanoLC-MS/MS ESI-Trap</td>
<td>MALDI-TOF/TOF</td>
<td>High resol nanoLC-MS/MS soon available</td>
<td>1000 / year</td>
<td>Turnaround time small-scale experiments</td>
</tr>
<tr>
<td>OncoProteomics Laboratory</td>
<td>NL</td>
<td>yes</td>
<td>we comply with CCKL</td>
<td>10 ml (platelet isolation)</td>
<td>Yes</td>
<td>nanoLC</td>
<td>tandem mass spectrometry</td>
<td>~4000-5000 nanoLC-MS/MS runs per year</td>
<td>Turnaround time small-scale experiments 4-8 weeks</td>
</tr>
<tr>
<td>Centre for Molecular Medicine and Biobanking</td>
<td>MT</td>
<td>no</td>
<td>no</td>
<td>no</td>
<td>western blotting</td>
<td>ELISA</td>
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This project has received funding from the European Union’s Horizon 2020 research and innovation programme under grant agreement No 676550.
Appendix III: Abbreviations

.map - A text file with no header file, and one line per variant with the following 3-4 fields: 1. Chromosome code 2. Variant identifier 3. Position in morgans/centimorgans 4. Base-pair coordinate

.ped – Original standard text format for sample pedigree information and genotype calls. Normally must be accompanied by a .map-file.

BAM – A binary format for storing sequence data

CEL – A data file created by Affymetrix DNA microarray image analysis software. Contains data extracted from probes on an Affymetrix GeneChip and can store thousands of data points.

CEN – European standardization organisation

CEN/TS – A Technical Specification is a normative document, used when various alternatives wouldn’t gather enough to allow agreement on a EN, but need to coexist in anticipation for future harmonization or for providing specifications in experimental circumstances/evolving technologies

CHP – File format that contains probe set analysis results generated from Affymetrix software

CNV - Word data conversion support file

EN – Standard confirmed by European standardisation organisation CEN

FASTQ - A text-based format for storing both a nucleotide sequence and its corresponding quality scores.

GLP – Good Laboratory Practice

IBBL PT – Proficiency Testing programme provided by the Integrated BioBank of Luxembourg

IDAT – A file format used to store BeadArray data from the myriad of genome wide profiling platforms on offer from Illumina Inc.

ISO – International Standardization Organization

IVT – In Vitro Transcriptomics

MS – Mass Spectrometry

NGS – Next Generation Sequencing

OD – Optical Density

QC – Quality Control

RIN – RNA Integrity Number

SNP – Single Nucleotide Polymorphism

TXT – Filename extension for text files

WP – Work Package

zCall - A variant caller specifically designed for calling rare single-nucleotide polymorphisms from array-based technology

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Appendix IV: Questionnaires

a) Genomics-questionnaire

Omics technology platform questionnaire – Genomics

We are gathering information on the omics technology service providers in Europe. The information will be used to improve the biobanks’ knowledge of available omics technology service providers.

We hope you have time to answer these questionnaires and we appreciate all your help and effort. This is a joint deliverable of ADOPT (H2020 project) Work Packages 2 and 6.

1. BACKGROUND INFORMATION

* Name: 
* E-mail: 
* Phone number: 
* Name of your biobank/facility: 
* Country: 
* Does your biobank/facility have an access: 

2. TECHNOLOGIES

Which technologies is your biobank/facility using?

- Affymetrix
- Illumina
- Other

If other, please specify: 

3. QUALITY

* Quality standard of your biobank/facility (ISO, CE, etc.): 
* Quality standard of the samples: 

4. REQUIRED DNA

* Quantity of DNA required: 
* Concentration of DNA required: 

Quality control is provided by your biobank/facility:

- Yes
- No

Other comments: 

5. DELIVERABLES

Deliverable throughout:

- Raw files including QAT files
- De-multiplexed fastq files
- Taxa files compatible with software such as Pilis
- Custom files
- CRV files

PROCEED

Submit

End of the survey, thank you for your time!

This project has received funding from the European Union’s Horizon 2020 research and innovation programme under grant agreement No 676550.
b) Transcriptomics-questionnaire

Omic technology platform questionnaire - Transcriptomics

We are gathering information of the omics technology service providers in Europe. The information will be used to improve the knowledge of omics technology service providers. We hope you have time to answer this short questionnaire and we appreciate all your help and effort. This is a short follow-up on ADOPTR0550-project (Transomics) Work Package 2 and 6.

1. BACKGROUND INFORMATION
   * Name:
   * Email:
   * Phone number:
   * Name of your biomedical facility:
   * Country:
   * Does your biomedical facility have open access:

2. TECHNOLOGIES

Which technologies do your biomedical facility use?
- Metagenomics
- Metromics
- Other

If other, please specify:

Throughput capacity of your lab and facility per month or per year:

3. QUALITY

* Quality standard of your biomedical facility (ISO, 218, etc.):
* Quality standard of the samples:

4. REQUIRED RNA

* Quantity of RNA required:
* Concentration of RNA required:

Quality control is provided by your biomedical facility:
- Yes
- No

Other comments:

5. DELIVERABLES

Deliverable throughput 1:
Deliverable throughput 2:
Deliverable throughput 3:
Deliverable throughput 4:
Deliverable throughput 5:

PROCEED

Submit

End of the survey, thank you for your time!

This project has received funding from the European Union’s Horizon 2020 research and innovation programme under grant agreement No 676550.
c) Metabolomics-questionnaire

Omics technology platform questionnaire - Metabolomics

We are gathering information of the omics technology providers in Europe. The information will be used to improve the biobanks' knowledge of possible omics technology services providers. We hope you have time to answer this short questionnaire and we appreciate all your help and effort. This is a part deliverable of ADOPTed project (Horizon 2020) Ward Packet 3 and 6.

1. BACKGROUND INFORMATION
   * Name:
   * E-mail:
   * Phone number:
   * Name of your biobank/facility:
   * Country:
   * Does your biobank/facility have open access:

2. TECHNOLOGIES
   Which technologies is your biobank/facility using?
   ○ NGS
   ○ MS
   ○ METABOLOMICS
   ○ Other
   If other, please specify:
   Throughput capacity of your biobank/facility (per month or per year):

3. QUALITY
   * Quality standard of your biobank/facility (ISO, CAP, etc.):
   * Quality standard of the samples:

4. REQUIRED SERUM/PLASMA
   * Quantity of serum/plasma required:
   Quality control is provided by your biobank/facility:
   ○ Yes
   ○ No
   Other comments:

5. DELIVERABLES
   Deliverable throughout 1:
   Deliverable throughout 2:
   Deliverable throughout 3:
   Deliverable throughout 4:
   Deliverable throughout 5:

6. PROCEED
   Submit

End of the survey, thank you for your time!

This project has received funding from the European Union’s Horizon 2020 research and innovation programme under grant agreement No 676550.
d) Proteomics-questionnaire

Omics technology platform questionnaire – Proteomics

We are gathering information of the omics technology service providers in Europe. The information will be used to improve the biobanks’ knowledge of
available omics technology service providers.

We hope you have time to answer the online questionnaire and we appreciate all your help and efforts. This is a part of the project EU project

1. BACKGROUND INFORMATION
   • Name:
   • E-mail:
   • Phone number:
   • Name of your biobank/facility:
   • Country:
   • Does your biobank/facility have access:

2. TECHNOLOGIES

Which technologies is your biobank/facility using?

Technology 1:
Technology 2:
Technology 3:
Technology 4:

Throughtput capacity of your biobank/facility (per month or per year):

3. QUALITY

• Quality standard of your biobank/facility (ISO, CRQ, ATP):
• Quality standard of the sample:

4. REQUIRED SERUM/PLASMA

• Quantity of serum/plasma required:

Other samples (tissue, biopsy, tissues) – specify sample types and quantity required:

Quality control is provided by your biobank/facility:
• Yes
• No

Other comments:

5. DELIVERABLES

Deliverable throughout 1:
Deliverable throughout 2:
Deliverable throughout 3:
Deliverable throughout 4:
Deliverable throughout 5:

PROCEED
Submit

End of the survey, thank you for your time!

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